CLINICAL SNAPSHOT

Gene Therapy Approaches to a Rare Retinal Disease: Choroideremia

A 28-year-old man reported reduced night vision and dark adaption since childhood, progressive visual field defects, glare, and at presentation, best-corrected visual acuity was reduced to 0.63 (ETDRS) in both eyes. Funduscopy revealed in both eyes a pale fundus with chorioretinal atrophy leading to exposure of the sclera and large choroidal vessels, widespread pigment clumping and a central island of relatively preserved retina. These clinical findings pointed to choroideremia, which was confirmed by multimodal retinal imaging, electrophysiological testing, and genetic testing. Choroideremia is a rare X-linked recessive retinal degeneration caused by mutations in the *CHM* gene. Comprehensive clinical characterization including molecular genetic testing is essential whenever choroideremia is suspected. Recent advances in the treatment of choroideremia include *in vivo* gene augmentation therapy. The disease is well suited for gene therapy approaches due to, amongst other factors, the accessibility of the eye and a large therapeutic window, with often early diagnosis and usually good visual acuity until the retinal degeneration involves the fovea. *In vivo* gene augmentation therapy is currently further explored in clinical trials of different stages.

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Conflict of interest statement: The authors have received third-party funding for the conduct of studies on behalf of Nightstar Therapeutics.

Translated from the original German by David Roseveare.

Cite this as: Birtel J, Holz FG, Herrmann P: Gene therapy approaches to a rare retinal disease: choroideremia. Dtsch Arztebl Int 2020; 117: 30. DOI: 10.3238/arztebl.2020.0030

Wide-angle false-color image of the left eye